Practitioners Manual
South Dakota
Newborn Screening

South Dakota
Department of Health
Newborn Screening Program
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INTRODUCTION

Newborn screening is a preventative public health surveillance program that is performed in every state in the United States and in many countries throughout the world. Newborn screening is a public health activity aimed at the early identification of infants who are affected by certain genetic/metabolic conditions. Early identification of these conditions is crucial as timely intervention can lead to a significant reduction of morbidity, mortality, and associated disabilities in affected infants.

Babies with these conditions may appear normal at birth. It is only with time that the biochemical abnormality affects the baby's health and development. By the time clinical symptoms appear, the damage may be permanent.

Newborn Screening in South Dakota
The South Dakota Department of Health is vested with the authority under SDCL 34-24-17 through 34-24-24 to assure all infants born in South Dakota are screened for metabolic disorders. A copy of the South Dakota laws and administrative rules pertaining to newborn screening can be found in the appendices of this manual (pages 30 - 37).

Purpose of the Manual
This manual will explain the necessary collaboration between the South Dakota Newborn Metabolic Screening Program (SDNMSP), the designated laboratory, healthcare facilities, and providers to help make newborn screening successful. Included is information about the disorders detected by the program and answers to frequently asked questions about newborn screening. This manual is intended to answer many of the questions health care providers generally have about the screening system. We hope that you will find this information helpful.

A Coordinated Effort
South Dakota's success in screening all newborns and providing appropriate follow-up for infants with abnormal tests or confirmed disorders depends on the coordinated efforts and involvement of many laboratory and health care provides. These include:

- SOUTH DAKOTA NEWBORN METABOLIC SCREENING PROGRAM (SDNMSP) which is responsible for statewide management of newborn screening, including program review and evaluation, follow-up and tracking of results, assessment, quality assurance, policy development, dissemination of educational brochures and manuals, developing and monitoring of the contract with the designated newborn screening testing laboratory, and promulgation of administrative rules.

- SOUTH DAKOTA PRACTITIONERS AND HOSPITALS who are responsible for the collection and handling of newborn screening specimens and prompt follow-up of abnormal results and unacceptable specimens. All infants must be screened prior to

hospital discharge. The primary care practitioners are responsible for providing parents with correct and current information and for prompt follow-up for abnormal screening results.

- SOUTH DAKOTA MEDICAL SPECIALISTS who, in conjunction with primary care providers, provide consultative and direct services regarding confirmatory testing, evaluation, treatment, and management of infants with disorders.

- SOUTH DAKOTA'S HOSPITAL AND CLINIC LABORATORIES, assist in the collection, handling, and shipping of specimens to the designated newborn screening testing laboratory.

- DESIGNATED CENTRALIZED NEWBORN SCREENING LABORATORY, the University of Iowa Hygienic Laboratory (UHL) in Ankeny, Iowa, is responsible for testing, record keeping, quality control of laboratory testing, and notification of test results.

- CHILDREN SPECIAL HEALTH SERVICES (CSHS - SOUTH DAKOTA DEPARTMENT OF HEALTH), provides care coordination, outreach clinics, and financial assistance with medical services for children diagnosed with the disorders detected through newborn screening for families who meet the CSHS eligibility guidelines.

WHEN TO COLLECT A BLOOD SPECIMEN

Full-term Infant/Hospital Birth setting

A filter paper specimen must be collected from each newborn infant as close as possible to the time of discharge and no later than 7 days of age by the institution or hospital where initial or subsequent newborn care was provided, regardless of prematurity, illness, feeding history or antibiotics given. The preferred time of collection is 24-48 hours of age.

Specimen Collected Early (< 24 hours)

If the initial specimen is collected before 24 hours of age, a second specimen must be collected within 2 weeks of age.

Premature/Sick Infants

A specimen should be collected as close as possible to discharge and no later than 7 days of life, unless a transfusion is imminent. The appropriate strategy is to ***always*** collect a newborn screening sample immediately **before any transfusions, regardless of the infant's age.**

Transfused Infant

Red blood cell (RBC) transfusions interfere with the interpretation of some newborn screening results. The appropriate strategy is to ***always*** collect a newborn screening sample immediately **before any transfusions, regardless of the infant's age.** Since red blood cells and plasma transfusions can cause false negative results, post-transfusion follow-up at the appropriate time is essential.

- Whenever possible, the newborn screen specimen should be collected prior to a transfusion of blood products, even if less than 24 hours of age.
- If the infant was transfused at the time of collection, a follow-up filter paper specimen must be collected at least 8 weeks after the last transfusion.

IMPORTANT: Always indicate on the specimen collection card if the infant has been transfused and document the date of the most recent transfusion.

Total Parenteral Nutrition (TPN Therapy - Hyperalimentation)

Infants on some types of total parenteral nutrition (TPN) may show elevated levels of amino acids (e.g. phenylalanine). Indications of TPN status on the collection form is necessary for clarifying some test results. The infant does not have to be off TPN before collecting the specimen.

The newborn screening test, like any laboratory test, may have false positives and false negatives. If signs and symptoms of one of the disorders are clinically evident, the physician should proceed to diagnostic testing, pending the results of the screening test or in spite of the results of the screening test.

Clinical Signs or Family History

There are a number of clinical situations that will modify the usual approach of obtaining a newborn screening specimen and waiting for the result. The following are brief suggested guidelines for particular situations that may arise in clinical practice. **Regardless of any diagnostic or therapeutic interventions, a newborn screen should be obtained on all infants to test for the other conditions included in the panel.** When in doubt about the course of management for any of the conditions on the screening test, consultation with a specialist is advised.

- **If the results of the newborn screen are pending:**
  For any of the screened conditions, but especially those in which the metabolite accumulation is dangerous, such as galactosemia, **treat as if the infant has the condition**. For other conditions, contact a metabolic center or metabolic physician for assistance with rapid diagnosis and institution of dietary treatment; for galactosemia, begin a soy-based formula until the screening results are known.
- **If the newborn screening test result was "normal":**
  If clinical symptoms suggest one of the screened conditions despite a "normal" screening test, the physician should **proceed as if the patient has the condition** and immediately contact a consultant specialist for instructions on further evaluation of the patient.

Newborn Screening of Infants with an Affected Close Relative

As many of the conditions tested for by newborn screening are genetic, it is possible that multiple members of a family may be affected. Prenatal diagnosis is possible for many of these conditions. If prenatal diagnosis determines that the infant is affected, any appropriate treatment (e.g., special diet) should be initiated immediately after birth.

If prenatal diagnosis predicts an unaffected baby, practitioners should bear in mind that no prenatal diagnostic test is 100% accurate. Neonates who are siblings or close relatives of an affected individual are not part of the "general population" for whom newborn screening is designed. For any infant with a positive family history, providers should contact appropriate consultant specialists, ideally prenatally or immediately at birth, to determine the proper diagnostic tests and proper timing of those tests.

Transferred Infants

When possible, the originating hospital should draw a newborn screening specimen **before** transferring the infant to another hospital. Transfer information should be included in the Certifier's Worksheet for Birth Certificate filing. The information will assist with follow-up if the newborn screening is not done prior to transfer.

Infants born in South Dakota must have testing performed by the designated laboratory. If an infant is transferred to another hospital before 48 hours of age, the receiving hospital must collect a specimen at an appropriate time within the first 48 hours of life. If an infant is transferred to another hospital after 48 hours of age, the transferring hospital must collect a specimen before the transfer and within the first 48 hours of age. When an infant born in South Dakota is transferred out of state, **the specimen needs to be submitted to the SD designated laboratory for testing**.

Home/Out-of Hospital Births

The parents, guardian, or custodian of each infant are responsible for having blood tests for metabolic disorders performed.

If a birth attendant was not present for the birth, the local registrar shall inform the parent or guardian of the need for a blood test for metabolic disorders when inquiring about or filing a certificate of birth.

Parent Refusal – What if the parents refuse the screen?

The form *"Parental Refusal of Newborn Screen"* is not a waiver. The form is a communication tool to the SDNMSP and to assist healthcare staff working with parents who refuse the metabolic screening. In the event an infant's parent, guardian, or custodian asks about refusing the newborn blood spot screening, ***strongly encourage testing*** and use the following procedure with the objective of obtaining parental consent. ***Only use the* "Parental Refusal of Newborn Screen" *form after exhausting all other reasonable alternatives*:**

1. Review the South Dakota Department of Health's "Newborn Screening" brochure with the parent, guardian or custodian. It is available for order from the Department of Health web site at www.state.sd.us/doh/Pubs/.

- Select "Order Online,"
- click on the "Family Health" tab,
- scroll to "Newborn Screening"
- choose NBS002 "Newborn Screening"

or copies can be also be downloaded from the Newborn Screening Program web site, www.state.sd.us/doh/NewbornScreening/links.htm.

2. Review the benefits of newborn blood spot screening and the risks of refusing this screening, to include the following:

Benefits: Early identification of children with the diseases included in newborn screening provides an opportunity to start treatment before symptoms appear. Early treatment for these diseases can prevent mental retardation, growth failure, severe illness and even an early death.

Risks of Refusing: If the child is not screened through the South Dakota Newborn Metabolic Screening Program, the child may have one of the diseases and will not receive the treatment necessary to prevent mental retardation, illness, or an early death. These diseases usually occur without a family history. Children with these

diseases often appear healthy at birth and may not have symptoms from the disease for several weeks or months.

3. Answer questions to ensure an informed decision; the SD Newborn Metabolic Screening Program can be contacted at (605) 773-3737 for assistance.

4. Review the "Parental Refusal of Newborn Screen" form with the parent(s).

5. Have parent(s) and witness sign the form; if they refuse to sign the form, document their refusal.

6. Retain the original form(s) or refusal documentation in the child's record.

7. Provide copies of the original to the parent(s), the child's physician, and the South Dakota Newborn Metabolic Screening Program. Mail or fax to:

South Dakota Department of Health
SDNMSP
615 East Fourth Street
Pierre, SD 57501-1700
Fax: (605) 773-5509

8. Provide documentation in the medical record who the copies were given to and the method of transmission, ie., in person, via mail, or by fax.

South Dakota Department of Health
Office of Family Health
Newborn Metabolic Screening Program
Parental Refusal of Newborn Screen

_____	_____
Name of Infant/child	Infant's Date of Birth
_____	_____
Full Name(s) of Infant's parent(s), guardian or custodian	Facility/Place of Infant's birth
_____	_____
Complete Address of Infant's parent(s), guardian or custodian	Phone no. for parent(s), guardian or custodian

The above-named infant's parent, guardian, or custodian, by signing below, hereby agrees that:

1. I have received and read the South Dakota Department of Health's "Newborn Screening" brochure regarding newborn screening tests for metabolic, endocrine, and hemoglobin disorders.

2. I have been informed and I understand that these tests are given to detect disorders that may not cause symptoms in my child for several weeks or months.

3. I have had explained to me and I understand the risks involved if I refuse to have my child screened.

4. I have been informed and I understand that if my child has one of the disorders to be screened, and the disorder is not detected due to my refusal to allow such testing, that delayed treatment of the disease/disorder may cause permanent damage to my child, including serious mental retardation, growth failure, and in some cases, death.

5. I have been informed that more information on newborn screening is available at: www.state.sd.us/doh/NewbornScreening/links.htm

I have discussed the testing requirements with _____
Hospital staff or witness
and I am refusing to allow my above-named child to be tested for these disorders.

_____	____/____/_____	_____
Signature	Date	Witness signature
_____		_____
Relationship to child		Witness (print name)

Original: Infant's Medical Record, provider copy

Copies: Parent
Infant's physician, and
South Dakota Newborn Metabolic Screening Program, mailed or fax to:
South Dakota Department of Health
SDNMSP
615 East Fourth Street
Pierre, SD 57501-1700 Fax: (605) 773-5509

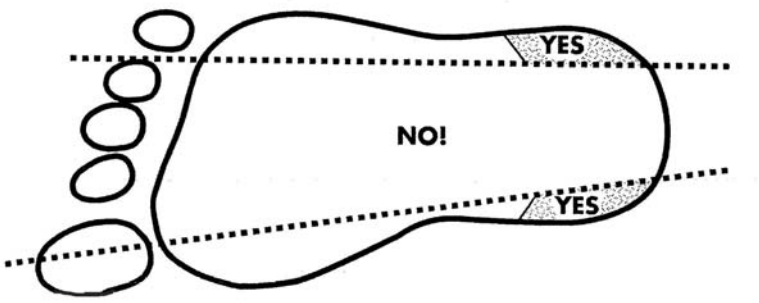
TECHNIQUES FOR BLOOD COLLECTION ON FILTER PAPER

Collection of dried blood spots on filter paper should be only by the direct heel stick procedure. Reference the National Committee on Clinical Laboratory Standards (NNCLS), now known as Clinical Laboratory Standards Institute (CLSI): LA4-A4 "Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard - Fourth Edition. To obtain copies of LA4-A4 contact CLSI at www.clsi.org or e-mail CustomerServcie@clsi.org or phone 610-688-0100.

The following instructions are consistent with the recommendations in *Blood Collection on Filter Paper for Neonatal Screening Programs*. National Committee for Clinical Laboratory Standards. Vol. 23 No. 21 Approved standard – 4th Edition, 2003 (NCCL Document LA4-A4).

Sampling Technique

Collect the blood onto the labeled filter paper, using the following protocol:



1. Cleanse infant's heel with 70% isopropyl alcohol (use only rubbing alcohol). *Note:* Warming the skin-puncture site with a warm moist cloth, or a heel warming device, for 3 minutes can increase blood flow through the site.

2. **Allow heel to *air dry*.**

3. Using a lancet, or heel incision device, and wearing gloves, perform the puncture on the plantar surface of the heel (as indicated in the drawing). The puncture should be made to a depth of less than 2.0 mm with a sterile lancet or incision device.

4. Gently wipe off first drop of blood with sterile gauze or cotton ball. The initial drop contains tissue fluids that may dilute sample.

5. **Wait for formation of large blood droplet; apply gentle pressure with thumb and ease intermittently as drops of blood form**.

6. Gently touch the printed side of the filter paper card to the blood drop and in one step, allow a sufficient quantity of blood to soak through and completely fill a pre-printed circle. **Do not** press the filter paper against the puncture site on the heel. Fill each printed circle with a SINGLE application of blood. Observe both sides of the filter paper card to assure that blood uniformly penetrated and saturated the card. Spotting should be done *only* on the printed side. The filter paper must not touch the skin puncture site.

7. Fill the required number of blood spots for mandated tests.

8. All used items should be disposed of in an appropriate biohazard container.

9. Elevate infant's foot above the body and apply pressure using sterile gauze. Do not apply adhesive bandages.

10. Allow blood specimen to AIR DRY THOROUGHLY, on a horizontally level—non-absorbent open surface, such as a plastic-coated test tube rack—**for a minimum of 3 hours** at ambient temperature and away from direct sunlight. Do not stack, heat, or allow to touch other surfaces during the drying process.

11. **Within 24 hours of collection**, prepare the dried blood collection card for shipment to the laboratory by courier or mail. Only use the mailing or courier envelope provided. Do not use plastic or plasticene envelopes. Humidity and moisture are detrimental to stability of dried blood spot specimens and can affect results.

Collection of NBS Specimen by Other Methods: Collection by Capillary Tube, Umbilical Arterial Catheter (UAC)

Mechanisms to communicate to the designated screening laboratory specimen collection by other methods are not in place. Cut-offs at the laboratory are based on assumption that the specimen was collected via direct heel stick procedure. Cutoffs have not been established for specimens collected by alternate methods such as capillary tube, dorsal hand vein or umbilical catheter.

MOST COMMON ERRORS IN SPECIMEN COLLECTION

INVALID SPECIMEN	POSSIBLE CAUSES
Quantity of blood not sufficient for testing (QNS)	Filter paper circles incompletely filled or not saturated/not all circles filled. Blood applied with needle or capillary tube. Contamination of surface of filter paper circle before or after specimen collection by gloved or ungloved hands, or by substance such as hand lotion or powder, etc.
Blood spots appear scratched or abraded	Blood applied improperly using capillary tube or other means (blotter has been damaged or torn by device).
Blood spots wet	Specimen not properly dried before mailing.
Blood spots appear supersaturated	Excess blood applied (usually with capillary tube or needle). Blood applied to both sides of filter paper.
Blood spots appear diluted, discolored, or contaminated	Puncture site squeezed or “milked.” Exposure of blood spots to direct heat. Contamination of filter paper before or after specimen collection by gloved or ungloved hands, or by substances such as alcohol, formula, water, powder, antiseptic solutions, or hand lotion. Contamination during transit.
Blood spots exhibit “serum rings”	Alcohol not wiped off puncture site before skin puncture is made. Filter paper has come into contact with alcohol, water, hand lotion, etc. Puncture site squeezed excessively. Specimen dried improperly. Blood applied to the filter paper with a capillary tube.
Blood spots appear clotted or layered	Same filter paper circle touched to a blood drop several times. Circle filled from both sides of the filter paper.
Blood will not elute from the blotter paper	Blood specimen has been heat-fixed. Blood specimen is too old (more than two weeks between collection and receipt by the screening laboratory).

COMPLETING THE SPECIMEN COLLECTION CARD

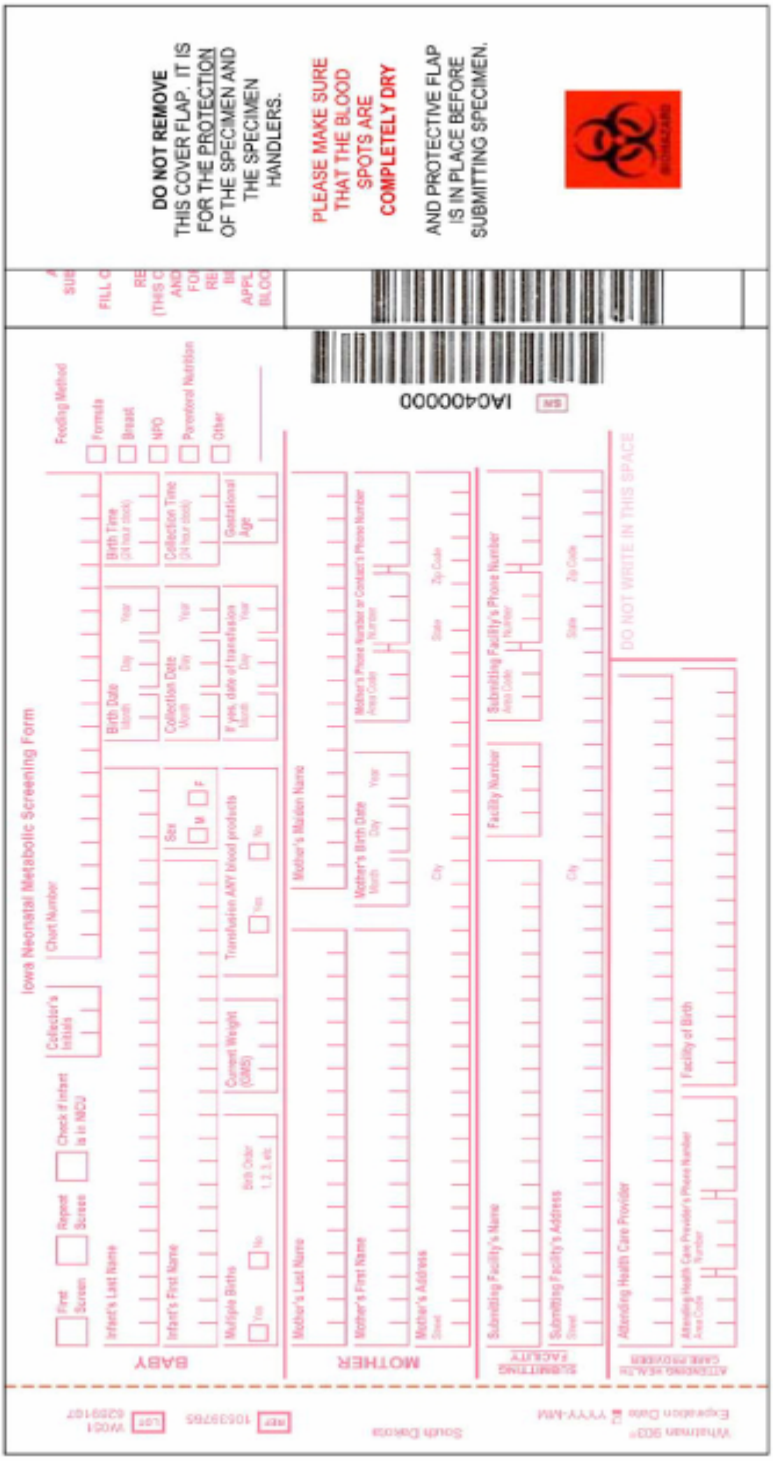
The lab specimen collection card is a legal record; the submitter is legally responsible for the accuracy and completion of all information.

All information requested is vitally important for the process of screening and follow-up. If key information is missing or unreadable, these specimens are difficult and time consuming to perform follow-up service; it also requires additional phone calls to the submitter to obtain the missing information. This may result in unnecessary delays in treating affected infants. It is extremely important that all requested information on the specimen collection card is filled out completely and **LEGIBLY**.

When filling out the specimen collection card,

- **Provide all information** to avoid having specimen rejected or test results withheld;
- **Use a ballpoint pen**, as soft-tip pens will not copy through to the carbon copies;
- **Use blue or black ink** and stay within the limits of the designated boxes;
- **Do not touch the filter paper,** as this could affect testing results;
- **Do not use plastic imprint cards**, as they produce unreadable information;
- **Do not use a typewriter to fill out the form**, as it may contaminate the filter paper;
- **Do not put labels or tape on the screening collection form or filter paper,** as it makes logging and tracking specimens in the lab very difficult.
- **Reattach the plies of the specimen collection card with a paper clip** if they become detached. Do not use adhesive tape.

South Dakota Newborn Screening Specimen Collection Card



The dried blood spot filter paper collection cards are three part forms. All demographic information must be completed, accurately and legibly before collecting the specimen. Every data field is critical to ensuring proper testing, and/or ensuring the ability to get the newborn back in if needed for repeat or confirmatory testing. Certain data elements are also critical to the matching done between the SDNMSP (metabolic testing) and the State Electronic Vital Records and Screening System (EVRSS -birth certificate).

Baby Information

First Screen/Repeat Screen - Mark if this is the first specimen collected for testing or if this is a repeat specimen.
Check if Infant is in NICU - Mark as appropriate.
Collector's Initials - Necessary for legal and quality assurance purposes.
Infant's Chart Number - This field is for the birth hospital or the collection facility's use. It is usually the newborn's assigned medical record number and may be helpful in tracking information and ensuring it goes with the right medical record.
Birth Date/Birth Time - Enter birth time using military time. This is one of the data elements essential to follow-up to ensure the right baby's information goes with the right baby's records. It is particularly important for multiple births to distinguish "baby A' from "baby B" and "baby C".
Collection Date/Collection Time - Time parameters are crucial to ensuring the baby is at least 24 hours of age. They are entered into the laboratory data system which calculates if less than 24 hours, and flags the specimen as needing a repeat.
Multiple Births - Mark as appropriate; use number as identified for infants of multiple births.
Current Weight - Transient elevations of 17-OHP, the analyte for congenital adrenal hyperplasia (CAH) screen may occur in pre-term and low birth weight babies. Because of this, four weight-related 17-OHP ranges are in place to minimize the number of false positive results. Without a weight indicated on the collection form, CAH results cannot be reported.
Transfusion ANY blood products - Do not leave this information field blank.
If yes, date of transfusion - Provide date of most recent transfusion.
Gestational Age - This information can be important in the interpretation of certain test results.

Mother Information

Mother's Last Name - Be sure to include the mother's maiden (birth) name along with the last and first names. **The mother's maiden name is a key component used by the SDNMSP to match birth certificates with the newborn screening test results.**

Submitting Facility

Submitting Facility's Name - The submitter listed on the Specimen Collection Card is the health care facility or provider that collected the specimen and will receive the results after screening.
Facility Number - This is the number provided to the submitting facility from the designated laboratory.

Submitting Facility's Phone Number - For the notification and reporting purposes of the designated laboratory.

Attending Health Care Provider

Attending Health Care Provider - The responsibility for follow-up of an abnormal result rests with the physician of record, as identified on the lab requisition. In some cases this physician will not be seeing the baby after discharge from the birth hospital. For this reason it is recommended that prior to discharge, the hospital ensure the medical record includes the name and address of the physician who will be caring for the child after discharge, and that they have contact information for the parent(s)/guardian. Rapid follow-up of an abnormal screening test depends upon identifying the physician who is caring for the child.

Attending Health Care Provider's Phone Number - For the notification procedures regarding abnormal test results.

Facility of Birth - The facility of birth is a required data element used by the SDNMSP to match birth certificates with the newborn screening test results. Be sure to provide this information even if it is the same as the submitting facility. If the infant was born outside of a South Dakota birth facility (ie. homebirth, or an out-of-state birth), please enter HOMEBIRTH or the abbreviation of the State the infant was born in (ie. MN, IA, ND, NE, or WY).

Feeding Method

Mark as appropriate. More than one box may be checked.

Unique Identification Number - Metabolic Screening Number (the number labels that are part of the specimen collection card)

It is important that all hospitals performing newborn screening utilize the sticker/label with the **unique identification number** on the Certifier's Worksheet for Completing the Birth Certificate. Each birth facility should have a process to place the unique identifier number (sticker) from the collection card on the Certifier's Worksheet for Completing the Birth Certificate. This is a critical step that allows the matching of the infant's metabolic screening results to their birth record.

HOW TO ORDER SUPPLIES

Collection forms, envelopes and NBS information pamphlets are available at no cost.

- **Specimen Collection Cards:** To order collection forms, contact University Hygienic Laboratory (UHL) at 515-725-1630 or by FAX at 515-725-1650.

- **Newborn Screening Pamphlet:** available at no charge. To order pamphlets call SDNMSP at 1-605-773-3737.

- **Practitioner's Manual - South Dakota Newborn Metabolic Screening**
  Available at no charge. Copies can be obtained by contacting the South Dakota Newborn Screening Program at 605-773-3737, or downloading at:

  http://www.state.sd.us/doh/NewbornScreening/links.htm

SPECIMEN TRANSPORT

Allow the blood specimen to air dry thoroughly (3-4 hours) on a level, non-absorbent surface. Insufficient drying can adversely affect the test results. Hair dryers, direct sunlight, or other sources of heat can not be used to dry the specimen.

As soon as possible within 24 hours of collection, place the dried specimen collection card inside the provided envelope and ship by UHL designated courier. For further instructions regarding courier pickup contact the UHL at (515) 725-1630 or (515) 725-1631.

LABORATORY TESTING AND REPORTING

Designated Laboratory
In order to conduct effective, timely follow-up for newborns affected by any of the disorders, the South Dakota Department of Health uses a centralized system to coordinate analyzing, reporting, and follow-up of newborn metabolic screens.

Beginning June 1, 2007, the University of Iowa Hygienic Laboratory (UHL) in Ankeny, Iowa, is the laboratory authorized to conduct newborn screening services for the State of South Dakota. **All newborn screening specimens for infants born in South Dakota must be submitted to UHL.** The UHL newborn screening laboratory operates every day including nights, weekends and holidays, 365 days/year. UHL is responsible for testing, record keeping, quality control of laboratory testing, and the notification of test results.

Notification of Newborn Screening Test Results
All providers are to ascertain the results of newborn screening on any infant in their care. Do not presume that a newborn screening test was obtained, or that the results of the newborn screen were normal. Laboratory reports will be sent to the submitter of the specimen. These results are to be used as a record for the child's medical chart.

Normal Test Results
Normal results in hard copy format will be sent to the submitter by USPS to be placed in the child's medical record.

Unacceptable Specimens - Poor Quality - Recollect
The UHL NBS Laboratory may consider a specimen to be of *poor quality* due to any of the following reasons: insufficient quality, layering, blood applied to both sides of the filter paper, blood not allowed to dry, contamination, serum separation, received more than 14 days after collection, etc. Before any specimen is rejected as "Poor Quality" two staff members examine it and agree to reject it (based on NCCLS document LA4-3A3). Please review *Techniques for Blood Collection on Filter Paper*, page 8 of this manual.

Since a poor quality specimen may compromise test results, submitters are notified immediately (with in 24 hours) by fax notification by UHL. UHL will identify these specimens by a "PQ" for Poor Quality. This notification indicates the need for a recollected specimen as soon as possible.

Invalid specimens, those with missing information, specimens collected after transfusion, and specimens collected from infants prior to 24 hours of age are tracked each workday and the submitting health care provider (and SDNMSP) will be notified by fax to collect another specimen as soon as possible.

Repeat Testing
As part of the contract with the designated newborn screening laboratory, follow-up staff are required to notify the submitter, physician and the SDNMSP of any specimen needing a repeat specimen collected on filter paper: early collection, unsatisfactory specimen, transfused specimen, and some abnormal or inconclusive results requiring only repeat filter paper specimen testing. The SDNMSP also communicates with the infant's physician to assure they have received notification of abnormal results and will monitor for repeat testing results.

Abnormal Test Results (Presumptive Positive)
As part of the contract with the designated newborn screening laboratory, follow-up staff are required to notify the submitter, physician and SDNMSP of any abnormal screening result. Some of these will fall in a "borderline" range requiring only a repeat filter paper specimen at that point in time. Others will require a different type of specimen to be tested by a different methodology.

Confirmatory Testing
Confirmatory testing information and instructions will be included with the notification process of presumptive positive results. Pay particular attention to the testing that is needed and the correct specimen type.

Link to the Laboratory Web Site
University of Iowa Hygienic Laboratory (UHL) in Ankeny, Iowa
http://www.uhl.uiowa.edu/services/mychildshealth/newborn/index.xml

DISORDERS SCREENED BY THE PROGRAM

These are disorders which may have significant mortality and morbidity when not diagnosed pre-symptomatically and may not be consistently identified clinically in the neonatal period. Early detection and treatment may improve the health and development of newborns identified with these disorders. More information on these conditions are available on the South Dakota Newborn Screening Website: http://www.state.sd.us/doh/NewbornScreening/

ENDOCRINE DISORDERS:

- Congenital adrenal hyperplasia (CAH) *
- Congenital hypothyroidism (CH) *

CYSTIC FIBROSIS *

HEMOGLOBINOPATHIES*:

- Sickle cell disease and other hemoglobin disorders

METABOLIC DISORDERS:

- Biotinidase deficiency *
- Galactosemia *

Amino Acid Disorders - Disorders identified through tandem mass spectrometry testing, listed with abbreviations and names:

- (ASA) Argininosuccinate acidemia*
- (CIT 1) Citrullinemia or ASA Synthetase Deficiency*
- (HCY) Homocystinuria (cystathionine beta synthetase)
- (MSUD) Maple Syrup Urine Disease*
- (PKU) Phenylketonuria*
- (TYR-1) Tyrosinemia Type 1*
- (ARG) Arginiemia**
- (BIOPT-BS) Defects of biopterin cofactor biosynthesis**
- (CIT-II) Citrullinemia type II**
- (BIOPT-RG) Defects of biopterin cofactor regeneration**
- (H-PHE) Benign hyperphenylalaninemia**
- (MET) Hypermethioninemia**
- (TYR II) Tyrosinemia type II**
- (TRY III) Tyrosinemia type III**

Fatty Acid Oxidation Disorders - Disorders identified through tandem mass spectrometry testing, listed with abbreviations and names:

- (CUD) Carnitine uptake defect (Carnitine transport defect)
- (LCHAD) Long-chain L-3 hydroxyacyl-CoA dehydrogenase*
- (MCAD) Medium chain acyl-CoA dehydrogenase*
- (TRP) Trifunctional protein deficiency*
- (VLCAD) Very long-chain acyl-CoA dehydrogenase*
- (CACT) Carnitine acylcarnitine translocase**
- (CPT-Ia) Carnitine palmitoyltransferase I**
- (CPT-II) Carnitine palmitoyltransferase II**
- (GA-II) Glutaric acidemia Type II**
- (MCKAT) Medium-chain ketoacyl-CoA thiolase**
- (M/SCHAD) Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase**
- (SCAD) Short-chain acyl-CoA dehydrogenase**

Organic Acid Disorders - Disorders identified through tandem mass spectrometry testing, listed with abbreviations and names:

- (GA-1) Glutaric acidemia type 1*
- (HMG) 3-Hydroxy 3-methylglutaric aciduria *
- (IVA) Isovaleric acidemia*
- (3-MCC) 3-Methylcrotonyl-CoA carboxylase*
- (Cbl-A,B) Methylmalonic acidemia (vitamin B12 disorders)*
- (BKT) Beta Ketothiolase*
- (MUT) Methylmalonic Acidemia (methylmalonyl-CoA mutase)*
- (PROP) Propionic acidemia*
- (MCD) Multiple carboxylase*
- (2M3HBA) 2-Methyl-3-hydroxybutyric aciduria**
- (2MGB) 2-Methylbutyrl-CoA dehydrogenase**
- (3MGA) 3-Methylglutaconic aciduria**
- (Cbl-C, D) Methylmalonic acidemia**
- (IBG) Isobutyrl-CoA dehydrogenase**
- (MAL) Malonic acidemia**

* American College of Medical Genetists Recommended Disorders - Core Panel

** American College of Medical Genetists Recommended Disorders - Secondary Targets

Caveat: The possibility of a false negative or a false positive result must always be considered when screening newborns for metabolic disorders.

Table I
Summarizes the disorders screened in South Dakota, including the incidence, symptoms, and treatment.

TABLE I

SUMMARY OF DISORDERS SCREENED BY THE PROGRAM

Condition	Compound Tested for	Incidence	Symptoms if Not Treated	Treatment
Endocrine Disorders:				
Congenital Adrenal Hyperplasia (CAH)	17-OH Progesterone	1:12,000	Addisonian Crisis in all infants; salt wasting in 2/3: dehydration, shock, hyperkalemia; virilization of females	Glucocorticoid and/or mineralocorticoids (Florinef)
Congenital Hypothyroidism	TSH	1:3,000	Mental retardation, other brain damage; growth delay	Thyroid hormone (L-Thyroxine)
Hemoglobin Disorders:				
Hemoglobinopathies including sickle cell anemia	Hemoglobin patterns	1:15,000 (1:400 in African Americans)	In sickle cell disease: death by sepsis or splenic sequestration anemia, sickling crises	Penicillin & comprehensive care
Metabolic Disorders:				
Biotinidase deficiency	Biotinidase	1:60,000	Mental retardation, seizures, skin rash, alopecia, hearing loss, death	Biotin
Galactosemia	Galactosemia enzyme (GALT)	1:60,000	Severe brain damage; liver disease; cataracts; death	Galactose-restricted diet
Amino Acids:				
Arginase Deficiency	Arginine	1:60,000	Irritability; develop-mental delay; spastic tetraplegia	Low protein diet, medication
Arginosuccinate Lyase Deficiency (ASA)	Arginine/Citrulline	1:60,000	Hyperammonemia; mental retardation' seizure; death	Low protein diet, medication
Citrullinemia	Citrulline	1:60,000	Hyperammonemia: mental retardation; seizure; death	Low protein diet, medication

TABLE I (continued)

SUMMARY OF DISORDERS SCREENED BY THE PROGRAM

Condition	Compound Tested for	Incidence	Symptoms if Not Treated	Treatment
Amino Acids: (continued):				
Homocystinuria	Methionine	1:100,000	Mental retardation; dislocation of lenses; marfanoid body habitus	Pyridoxine: methionine restricted, cysteine supplemented diet
Hyperphenylal-aninemia, including phenylketonuria	Phenylalanine	1:10,000	Profound mental retardation; seizures	Low phenylalanine diet
Tyrosinemia (type II may not be detected on NBS)	Tyrosine	1:100,000	Vomiting, lethargy; liver disease; coagulopathy renal tubular acidosis	Medication; low phenylalanine/low tyrosine diet
Organic Acidemias: • Beta-ketothiolase deficiency • Glutaric acidemia, Type I • Isobutyryl CoA dehydro-genase deficiency • Isovaleric acidemia • Malonic aciduria • Maple Syrup Urine Disease (MSUD) • Propionic acidemia • 3-Hydroxy-3-methylglutaryl (HMG) CoA lyase deficiency • 2-Methyl-3-hydroxybutyryl CoA dehydrogenase deficiency • 3-Methylcrotonyl CoA carboxylase deficiency	Acylcarnitines	1:53,000	Neonatal onset: irritability; lethargy; ketoacidosis, coma; death Late onset; failure to thrive; hypotonia; mental retardation Some will be asymptomatic	Dietary therapy/ Medications

TABLE I (continued)

SUMMARY OF DISORDERS SCREENED BY THE PROGRAM

Condition	Compound Tested for	Incidence	Symptoms if Not Treated	Treatment
Fatty Acid Oxidation Defects: • Carnitine uptake/ transport defects • Multiple acyl-CoA dehydrogenase deficiency (MADD) • Short chain acyl-CoA dehydrogenase deficiency (SCAD) • Medium chain acyl-CoA dehydrogenase deficiency (MCAD) • Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD) • Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)	Acylcarnitines	1:9,300	"Reyes Like" episodes; hypoketotic hypoglycemia; lethargy; cardiomyopathy; hypotonia; mental retardation; coma; death Mother may have had AFLP/HELLP syndrome; acute fatty liver of pregnancy	Dietary therapy/ Medications

RESOURCES

SDNMSP Phone Numbers

South Dakota Newborn Metabolic Screening Program (SDNMSP).....1-605-773-3737
..... or 1-800-738-2301

Website Address

SDNMSP Web page .....http://www.state.sd.us/doh/NewbornScreening

Publications

Pamphlet "Newborn Screening" available at no charge. To order pamphlets call SDNMSP at 1-605-773-3737

Manual

"Practitioners Manual South Dakota Newborn Metabolic Screening", May 2007.
Copies are available to be downloaded from the SDNMSP web page
http://www.state.sd.us/doh/NewbornScreening/links.htm
or call the SDNMSP at 1-800-738-2301 or 1-605-773-3737.

SDNMSP Medical Consultants 1-605-333-7188

- Laura Davis-Keppen, MD - Pediatric Endocrinology, Genetics & Metabolic Disease
- James Wallace, MD - Pediatric Pulmonology (Cystic Fibrosis)
- Linda Stout, MD - Pediatric Hematology (Hemoglobinopathies)

Designated Laboratory……..University Hygienic Laboratory, Ankeny IA

Program Manager, Stan Berberich……..319-335-4500
Marcia Valbracht, Lab Supervisor.....515-725-1631
University Hygienic Laboratory Website:
http://www.uhl.uiowa.edu/

Video

NCCLS (National Committee for Clinical Laboartory Standards) Educational Video Tape and Manual, *Making a Difference Through Newborn Screening: Blood Collection on Filter Paper.* This video is available for loan. Contact: University Hygienic Laboratory (UHL) at 515-725-1630. This is an excellent resource for OB, nursery and laboratory personnel.

Related Websites

American Academy of Pediatrics
http://www.aap.org/

American Academy of Pediatrics Screening Fact Sheets
http://aappolicy.aappublications.org/

Center for Disease Control and Prevention
http://www.cdc.gov/genomics/activities/newborn.htm

American College of Medical Genetics (ACMG)
Newborn Screening ACT Sheets and Confirmatory Algorithms
http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

Genetics Home Reference - Provides consumer-friendly information about the effects of genetic variations on human health.
http://ghr.nlm.nih.gov/

National Newborn Screening and Genetics Resource Center (NNSGRC) - Provides information and resources in the area of newborn screening and genetics to benefit health professionals, the public health community, consumers and government officials.
http://genes-r-us.uthscsa.edu/

Heartland Regional Genetics and Newborn Screening Collaborative is a network of genetics and newborn screening providers, advocates and other stake holders from Arkansas, Iowa, Kansas, Missouri, Nebraska, North Dakota, Oklahoma and South Dakota.
http://heartland.ouhsc.edu/

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APPENDICES

Glossary of Terms and Acronyms

1. Attending health care provider - the licensed physician, nurse practitioner, certified midwife or physician assistant providing care to an infant at birth.

2. Birth attendant – a person licensed or certified by the state to provide maternity care and to deliver pregnant women.

3. Central laboratory – the designated laboratory.

4. CH – congenital hypothyroidism.

5. Confirmatory testing - test/s to prove or disprove the presence of a specific condition identified by screening tests. This testing is from a specimen other than the screening specimen.

6. Days of age – the measure of the age of a newborn in 24-hour periods; i.e., a newborn is one day of age 24 hours following the hour of birth.

7. Department – the South Dakota Department of Health.

8. Designated laboratory – a laboratory or laboratories authorized by the department pursuant to 44:19:02:03.01 to perform newborn screening services for the residents of South Dakota.

9. Discharge – the release of a newborn from the care of an institution or hospital to the parents or into the community.

10. Enzyme – a complex organic compound secreted by living cells which causes or accelerates chemical reactions or changes in a substrate.

11. EVRSS – the South Dakota Electronic Vital Records and Screening System that links newborn metabolic screening and newborn hearing screening with each infant's birth certificate.

12. Expanded screening panel - analysis of acylcarnitine and amino acids using tandem mass spectrometry (MS/MS).

13. Follow-up specimen – a second specimen collected from a newborn because the test results on a previously collected specimen were inconclusive or abnormal.

14. Follow-up test – a second specimen collected because the previous specimen or test results were inconclusive or abnormal.

15. FT4 – free thyroxine.

16. Galactosemia – a metabolic disease indicated by the presence of an excessive amount of galactose in the blood due to a deficiency of galactose-1-phosphate uridyl transferase.

17. Hypothyroidism – a disease indicated by low level of thyroxine in the serum of the newborn.

18. Inadequate specimen – a newborn's blood specimen which is not suitable in quality or quantity for performing newborn screening for one or more of the disorders requested.

19. Indeterminate result - test results do not adhere to present criteria established for interpretation of a normal or an abnormal result. Additional testing usually recommended.

20. Initial abnormal specimen – a newborn's first blood specimen which is defined as positive for reporting purposes.

21. Initial specimen – the first screening specimen collected subsequent to birth.

22. Initial test – the first valid screening test or combination of tests of a newborn for each disorder.

23. Metabolic disease – a malfunction in the processes that convert compounds to protoplasm, energy, and waste.

24. Newborn – an infant 28 days of age or under.

25. Newborn's physician – the physician responsible for the care of a newborn after discharge from the hospital.

26. Phe – blood phenylalanine.

27. PKU "phenylketonuria" – a metabolic disease indicated by an excessive amount of phenylalanine in the serum of the newborn.

28. Poor quality specimen - insufficient quantity, layering, blood applied to both sides of the filter paper, blood not allowed to dry, contamination, serum separation, received more than 14 days after collection.

29. Premature infant - infant born before completing week 37 of gestation.

30. Presumptive positive - abnormal or positive newborn screening test results

31. Tandem mass spectrometry - is an analytical technique for identifying compounds based on their mass-to-charge ratio.

32. T4 – symbol for thyroxine.

33. TPN – total parenteral nutrition or hyperalimentation.

34. Transfer – the release of a newborn from the care of one institution or hospital to the care of another institution or hospital.

35. TSH – thyroid stimulating hormone.

36. Unacceptable or unsatisfactory specimen – a newborn's blood specimen that is not suitable in quality or quantity for performing newborn screening for one or more of the disorders covered by this manual.

SDNMSP BLOOD COLLECTION SUPPLIES

All newborn screening blood collection supplies are available from the designated laboratory (UHL) at no charge by calling:

To order specimen collection cards, contact University Hygienic Laboratory (UHL) at 515-725-1630 or by FAX at 515-725-1650.

South Dakota Newborn Metabolic Statutes

34-24-17. Screening of newborn infants for metabolic disease. All infants born in the state of South Dakota shall be screened for metabolic disease. This screening shall be as prescribed by the state department of health.

34-24-18. Phenylketonuria, hypothyroidism and galactosemia testing in newborn. The tests for detecting metabolic disorders of the newborn infant, as prescribed by the department of health, shall include, but not be limited to, the testing for excessive phenylalanine in the serum of the newborn, for hypothyroidism and for elevated blood galactose in the newborn.

34-24-19. Phenylketonuria, hypothyroidism or galactosemia tests when facilities not available. If facilities are not available for the screening of newborn infants for the Phenylketonuria syndrome, for hypothyroidism or for galactosemia, the department of health shall arrange for testing through the director of laboratories.

34-24-20. Phenylketonuria, hypothyroidism and galactosemia tests provided by department for newborn not tested. If the required report to the department of health shows that the newborn infant was not tested for Phenylketonuria, for hypothyroidism or for galactosemia, the department may arrange for the infant to be tested.

34-24-21. Procedures prescribed after positive Phenylketonuria, hypothyroidism or galactosemia tests. If a screening test indicates a newborn infant may be afflicted with the Phenylketonuria syndrome, hypothyroidism or galactosemia, the department of health shall prescribe the procedures to be followed in order to determine if the syndrome is actually present.

34-24-22. Testing for other metabolic diseases. When tests for detecting a metabolic disease other than Phenylketonuria, hypothyroidism and galactosemia are perfected, the department of health may require that tests for the syndrome or syndromes be made and reported to the health department.

34-24-23. Reports to department on metabolic disease tests -- Forms. Results of such tests for metabolic disorders in infants, as prescribed by the department of health, shall be sent to the department on forms to be prescribed and furnished by the department to all physicians, public health nurses and hospitals.

34-24-24. Follow-up on children with metabolic disease. It shall be the responsibility of the department of health to follow the development of all children carrying the syndrome of any metabolic disease to ensure that those persons responsible for the care of the child are fully informed of accepted medical procedures for the detection, prevention, and treatment of such condition.

34-24-25. Rules and regulations. The department of health is authorized to promulgate and enforce rules and regulations to aid in implementing the provisions of § 34-24-16 to 34-24-24, inclusive.

ARTICLE 44:19

NEWBORN SCREENING

CHAPTER 44:19:01

DEFINITIONS

44:19:01:01. Definitions. Terms used in this article mean:

(1) "Abnormal specimen," a newborn's blood specimen that is defined as positive for reporting purposes;

(2) "Birth attendant," a person licensed or certified by the state under SDCL title 36 to provide maternity care and to deliver pregnant women;

(3) "Days of age," the measurement of the age of a newborn in 24-hour periods; i.e., a newborn is one day of age 24 hours following the hour of birth;

(4) "Department," the South Dakota Department of Health;

(5) "Designated laboratory," a laboratory or laboratories authorized by the department pursuant to § 44:19:02:03.01 to perform newborn screening services for infants born in South Dakota;

(6) "Filter paper specimen," a sample of blood collected from an infant by saturating marked areas of a filter paper card with blood obtained by skin puncture of the heel;

(7) "Discharge," the release of a newborn from the care of an institution or hospital to the parents or into the community;

(8) "Initial specimen," the first metabolic screening specimen collected subsequent to birth;

(9) "Initial test," the first valid metabolic screening test or combination of tests of a newborn for each disorder;

(10) "Metabolic disorder," a malfunction in the processes that convert compounds to protoplasm, energy, and waste;

(11) "Newborn," an infant 28 days of age or under;

(12) "Newborn's physician," the physician responsible for the care of a newborn;

(13) "Newborn screening," a laboratory test performed on newborn blood specimens to identify newborns with metabolic disorders;

(14) "Repeat specimen," a specimen collected from a newborn because a previously collected specimen was either unsatisfactory or the test results were inconclusive, abnormal, or positive;

(15) "Repeat test," a test repeated because the previous specimen or test results were unsatisfactory, test results were not complete, or the test results were abnormal;

(16) "Secretary," the secretary of the department of health;

(17) "Submitter," the person who sends the newborn screening requisition form to the designated laboratory for initial, repeat, or confirmatory screening tests, including the hospital, laboratory, or physician;

(18) "Transfer," the release of a newborn from the care of one institution or hospital to the care of another institution or hospital;

(19) "TSH," thyroid stimulating hormone; and

(20) "Unacceptable or unsatisfactory specimen," a newborn's blood specimen that is not suitable in quality or quantity for performing newborn screening for one or more of the disorders covered by this article.

Source: 12 SDR 167, effective April 20, 1986; 15 SDR 59, effective October 19, 1988; 18 SDR 67, effective October 16, 1991; 23 SDR 91, effective December 9, 1996; 31 SDR 164, effective May 9, 2005.

General Authority: SDCL 34-24-25.

Law Implemented: SDCL 34-24-23.

44:19:01:02. Transferred to § 44:19:02:03.

44:19:01:03. Deadlines for reporting test results. Repealed.

Source: 12 SDR 167, effective April 20, 1986; repealed, 15 SDR 59, effective October 19, 1988.

44:19:01:04. Required tests. In addition to those metabolic disorders required in SDCL 34-24-18, any infant born in South Dakota shall be screened for the following metabolic disorders by a laboratory designated by the department to perform such testing:

(1) Hemoglobinopathies;
(2) Biotinidase deficiency;
(3) Classical congenital adrenal hyperplasia;
(4) Fatty acid oxidation disorders;
(5) Amino acid disorders; and
(6) Organic acid disorders.

* For changes effective May 7, 2007, check website at
http://legis.state.sd.us/rules/DisplayRule.aspx?Rule=44:19

Source: 31 SDR 164, effective May 9, 2005.
General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-22.

CHAPTER 44:19:02

NEWBORN SCREENING LABORATORY

44:19:02:01. Newborn screening laboratory test requisition. Repealed.

Source: 15 SDR 59, effective October 19, 1988; repealed, 23 SDR 91, effective December 9, 1996.

44:19:02:02. Standards for laboratory operation. Repealed.

Source: 15 SDR 59, effective October 19, 1988; 18 SDR 67, effective October 16, 1991; repealed, 23 SDR 91, effective December 9, 1996.

44:19:02:03. Reporting requirements. Repealed.

Source: 12 SDR 167, effective April 20, 1986; transferred from § 44:19:01:02, 15 SDR 59, effective October 19, 1988; 18 SDR 67, effective October 16, 1991; repealed, 23 SDR 91, effective December 9, 1996.

44:19:02:03.01. Designation of laboratories. In accordance with criteria in § 44:19:02:03.02 the department shall designate the laboratories that are authorized to perform newborn screening services for infants born in of South Dakota. The number of laboratories to be designated at any one time is at the discretion of the secretary.

Source: 23 SDR 91, effective December 9, 1996; 31 SDR 164, effective May 9, 2005.
General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-25.

44:19:02:03.02. Criteria for designation of laboratories. The department shall use the following criteria in designating laboratories to conduct newborn screening tests:

(1) Professional qualifications necessary for satisfactory performance of the required services;
(2) Demonstrated experience and technical expertise;
(3) Demonstrated capacity to accomplish work in the required time and maintain a continuity of services;
(4) Past performance on similar contracts with publicly or privately funded programs in terms of cost control, quality of work, and compliance with performance schedules;
(5) Proposed fee to be charged per specimen for newborn screening; and
(6) Any other criteria as deemed appropriate by the secretary.

Source: 23 SDR 91, effective December 9, 1996.
General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-25.

44:19:02:04. Public notice of designated laboratories. The department shall publish the names and addresses of the designated laboratories authorized to conduct newborn screening tests.

Source: 15 SDR 59, effective October 19, 1988; 23 SDR 91, effective December 9, 1996.

General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-25.

44:19:02:04.01. Designated laboratory responsibilities. The designated laboratory shall provide the following notices regarding specimens received:

(1) Any unacceptable or unsatisfactory specimen and the need for a repeat specimen shall be reported to the submitter no later than 24 hours after the analysis or within four working days of specimen receipt, whichever is earlier; and

(2) Any abnormal and positive test results shall be reported to the submitter no later than 24 hours after the analysis or within four working days after specimen receipt, whichever is earlier.

Source: 31 SDR 164, effective May 9, 2005
General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-17, 34-24-23, 34-24-24.

44:19:02:05. Responsibilities of parents. The parents, guardian, or custodian of each infant is responsible for having blood tests for metabolic disorders performed within the first 48 hours of an infant's life. If a parent, guardian, or custodian refuses to have a newborn tested for metabolic disorders, despite having been notified of the need for testing, the parent, guardian, or custodian shall sign a written statement regarding the refusal.

Source: 18 SDR 67, effective October 16, 1991; 23 SDR 91, effective December 9, 1996; 31 SDR 164, effective May 9, 2005.
General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-17, 34-24-23, 34-24-24.

44:19:02:06. Responsibilities of hospitals, physicians, and other health professionals. The attending physician, other health professional, hospital, or public health facility shall notify the parents, guardian, or custodian of each infant of the responsibility and need to have the newborn screening tests performed. The attending physician or other health professional shall place all newborn screening test results in the newborn patient's record. If a parent, guardian, or custodian refuses to have the newborn tested, the attending physician, other health professional, hospital, or public health facility shall obtain a written signed statement from the parent, guardian, or custodian of the infant regarding the refusal and place it in the newborn patient's record, notify the department within 24 hours of the refusal at 1-800-738-2301, and send a copy of the signed refusal to the department. If a parent, guardian, or custodian refuses to sign the statement, the attending physician, other health professional, hospital, or public health facility shall document such refusal, place it in the newborn patient's record, and send a copy of such documentation to the department.

Once the physician receives the results of the newborn screening test, the physician shall place the results in the newborn's medical record.

Source: 31 SDR 164, effective May 9, 2005.
General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-17, 34-24-23, 34-24-24.

44:19:02:07. Responsibilities of Department of Health. Upon notification by a designated laboratory of a positive screening result for an infant, the department shall work with the newborn's physician to locate the infant and facilitate the entry of the infant into further diagnostic and medical management services.

Source: 31 SDR 164, effective May 9, 2005.
General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-17, 34-24-23, 34-24-24.

CHAPTER 44:19:03

TIME SEQUENCE FOR TESTING

Section

44:19:03:01. Specimen submission to designated laboratory. Specimens must be submitted to a designated laboratory. Only filter paper specimens may be submitted for newborn screening.

Source: 15 SDR 59, effective October 19, 1988; 18 SDR 67, effective October 16, 1991; 23 SDR 91, effective December 9, 1996.
General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-18.

44:19:03:02. Collection of filter paper specimens. Filter paper specimens for submission to designated laboratories must be collected as follows:

(1) A filter paper specimen must be collected from each newborn infant as close as possible to the time of discharge and no later than seven days by the institution or hospital where initial or subsequent newborn care was provided regardless of prematurity, illness, feeding history, or antibiotics being given. The designated laboratory shall retain the specimen until the laboratory determines that the specimen can be destroyed;

(2) Filter paper specimens must be collected from newborns prior to a transfusion of whole blood, plasma, or packed red blood cells regardless of the infant's age. If the pre-transfusion specimen was collected before 24 hours of age, a repeat specimen must be collected 72 hours after the last transfusion. If the initial specimen was collected after a transfusion and there is no pre-transfusion specimen, a second specimen shall be collected at 90 days from the last transfusion; * For changes effective May 7, 2007, check website at **http://legis.state.sd.us/rules/DisplayRule.aspx?Rule=44:19**

(3) The receiving institution shall obtain filter paper specimens from newborns transferred within the first 48 hours of life;

(4) The institution initiating the transfer shall obtain blood specimens from newborns transferred after the first 48 hours of life;

(5) If a birth occurs that is not attended by a birth attendant, the local registrar shall inform the parent or guardian of the need for a blood test for metabolic disorders when inquiring about or filing a certificate of birth. The registrar shall inform the parent or guardian where the blood sample may be drawn or refer them to the local community health nurse; and

(6) For any specimen collected on a newborn less than 24 hours of age, a repeat specimen must be collected within two weeks.

Source: 15 SDR 59, effective October 19, 1988; 23 SDR 91, effective December 9, 1996; 31 SDR 164, effective May 9, 2005.
General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-18.

CHAPTER 44:19:04

CONSULTATION

Section
44:19:04:01 Consultation services.

44:19:04:01. Consultation services. The department shall contract with a consulting physician to provide interpretation of test results and consultation to the attending physician on request.

Source: 15 SDR 59, effective October 19, 1988; 23 SDR 91, effective December 9, 1996.
General Authority: SDCL 34-24-25.
Law Implemented: SDCL 34-24-21, 34-24-24.